t(11;21)(q21;q22)

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Published in Atlas Database: October 2011

Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t1121q21q22ID1592.html

DOI: 10.4267/2042/47287

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### Identity

**Note**

This translocation is different from the t(11;21)(q12;q22) with MACROD1/RUNX1 involvement.

### Clinics and pathology

**Disease**

Acute myeloid leukemia (AML)

**Epidemiology**

One case to date, a 65-year-old male patient with M2-AML (Dai et al., 2007).

**Evolution**

The patient died 10 months after diagnosis.

### Genes involved and proteins

**LPXN**

**Protein**

LPXN contains two types of protein-protein interaction domains: leucine-aspartate (LD) repeats in N-term, and LIM (Lin-11 Isl-1 Mec-3) domains at the C-term. Belongs to the paxillin family (PXN, LPXN, TGFB1I1). Protein involved in focal adhesion. LPXN and paxillin had opposite roles in adhesion to collagen LPXN siRNA stimulated whereas paxillin siRNA inhibited cell adhesion. Strongly expressed in hematopoietic cells. LPXN is involved in bone resorption and stimulates prostate cancer cell migration (Chen and Kroog, 2010).

**RUNX1**

**Location**

21q22.3

**Protein**

Transcription factor (activator) for various hematopoietic-specific genes.

### Result of the chromosomal anomaly

**Hybrid gene**

**Description**

5' RUNX1 - 3' LPXN

**Transcript**

Two in frame fusion transcripts -fusion of exon 5 or 6 of RUNX1 to LPXN exon 8.

**Fusion protein**

**Description**

The two variant fusion proteins RUNX1-LPXN localized in the nucleus and inhibited RUNX1 transactivation (Dai et al., 2009). It is hypothesized that the reciprocal LPXN-RUNX1 may also play a role in leukemogenesis.
References


Chen PW, Kroog GS. Leupaxin is similar to paxillin in focal adhesion targeting and tyrosine phosphorylation but has distinct roles in cell adhesion and spreading. Cell Adh Migr. 2010 Oct-Dec;4(4):527-40.

This article should be referenced as such: